Familial dyskeratotic comedones: A rare entity

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ABSTRACT

Familial dyskeratotic comedones is an inherited disorder with distinctive clinical features and a disease course that is refractory to treatment. It is clinically characterized by numerous, discrete, disseminate, hyperkeratotic papules and comedones. On histopathology, it shows crater-like invaginations filled with keratinous material and evidence of dyskeratosis. We report here one family from central India with this rare disorder.

Key words: Comedones, dyskeratosis, familial comedones

INTRODUCTION

Familial dyskeratotic comedones (FDC) is a rare autosomal dominant condition. Rodin *et al*.[1] first reported this condition in 1967, and since then scanty cases were reported in literature. It is clinically characterized by numerous, discrete, disseminate, hyperkeratotic papules, and comedones.[1-10] On histopathology, it shows crater-like invaginations filled with keratinous material and evidence of dyskeratosis.[4-10] To the best of our knowledge, only 15 patients from seven families have been reported in the literature.[4]

CASE REPORT

A 32-year-old woman presented with multiple, small, asymptomatic black lesions predominantly over face, both limbs, and the trunk. Lesions first appeared over both the legs when she was 13- years-old. They gradually increased in number and subsequently spread to involve all areas of the body except the scalp, mucosa, palms, and the soles. Till the recent past, she had exacerbations and remissions. There was a family history of similar lesions; they had appeared around the age of 12 years in her mother, two older sisters, maternal uncle, and the patient's 14-year-old son and 9-year-old daughter [Figure 1]. There was no history of consanguinity among her parents.

Cutaneous examination showed numerous, widespread, hyperkeratotic papules and comedones varying in size from 0.5 to 1 cm over all areas of the body, especially the face, back, flexor aspects of arms along with multiple pock-like scars on the face and prominently over the nose [Figure 2]. The scalp, mucosa, palms and soles were free of lesions. Few of the lesions were larger in size from which keratinous material was expressed. There was no evidence of acne. Systemic examination and routine hematological investigations were normal.Histopathology revealed hyperkeratosis, parakeratosis and multiple crater-like invaginations containing keratinous material at places. No acantholysis was seen. There was irregular thinning of the epidermis and dyskeratosis was observed at places [Figure 3]. A mild chronic inflammatory infiltrate was seen in upper dermis with melanin.

Her daughter showed numerous, widespread, hyperkeratotic papules, and comedones with a predisposition for the limbs [Figure 4]. Biopsy could not be drawn from her daughter as parents declined consent.

DISCUSSION

FDC is an autosomal dominant disorder[7-10] with the following distinctive features:[2]

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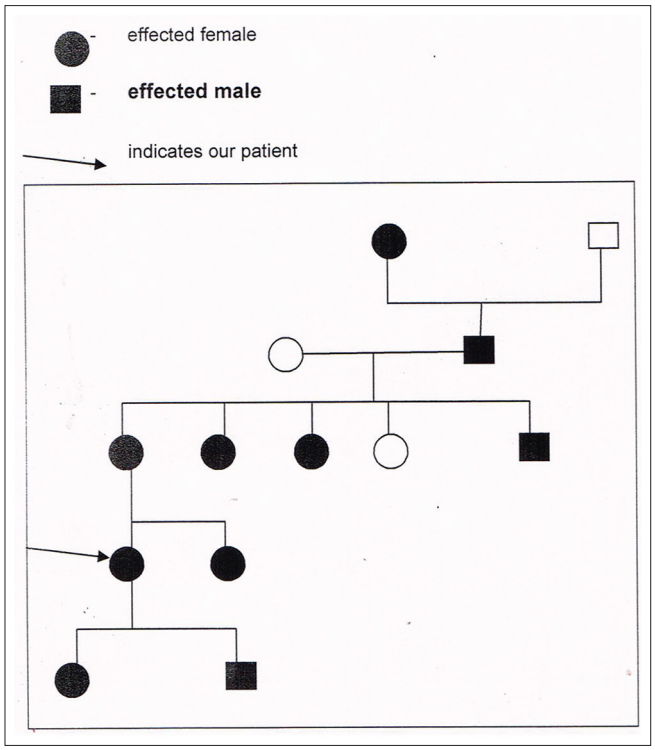

Figure 1: Pedigree chart

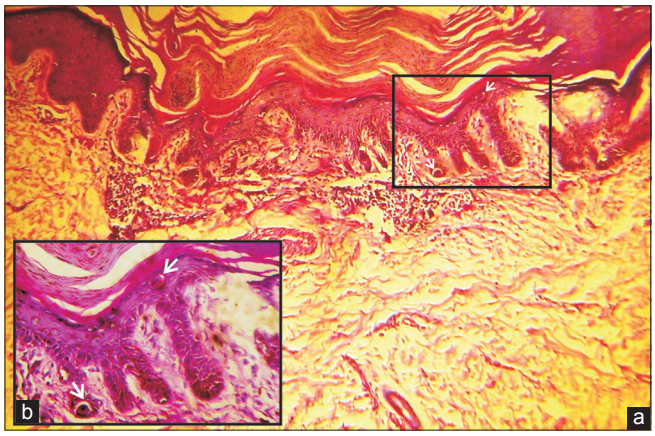

Figure 3: (a) Microphotograph showing massive hyperkeratosis, parakeratosis, and epidermal invagination with keratinous material (H and E; original magnification ×40) (b) Showing enlarged view of marked portion in Figure 2a) showing dyskeratotic cells shown by arrows (H and E; original magnification ×100)

1. Lesions clinically resembling comedones
2. Occurrence in some family members
3. Presence of dyskeratotic changes on histological examination.

The lesions start as pinpoint dark papules with predilection for the trunk, arms, legs, face and shaft of the penis (sparing the glans), oral mucosa, palms, and soles.[6] It usually presents at an adolescent age, but can present as dark papules even at a younger age.[10] Classical lesions are not associated with acne, but it may coexist.[3,4,6] Although the condition is mostly

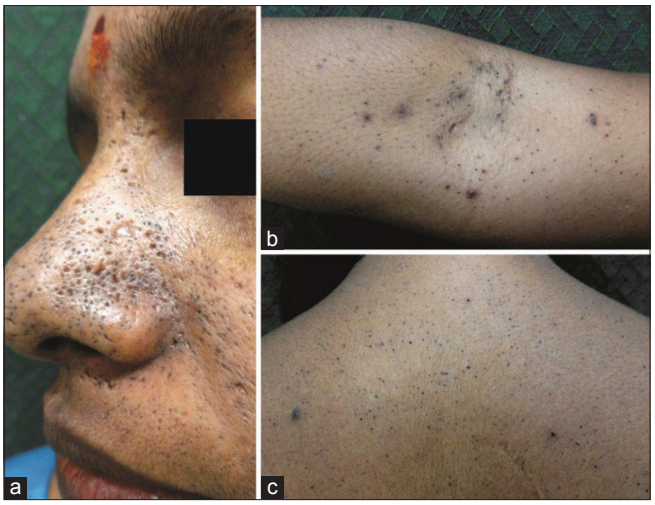

Figure 2: (a) Showing multiple, deep, pock-like scars with comedones over nose and cheek (b) Multiple comedones with pigmented papular lesions over flexor aspect of arm (c) Multiple comedones and pigmented papules on the upper back

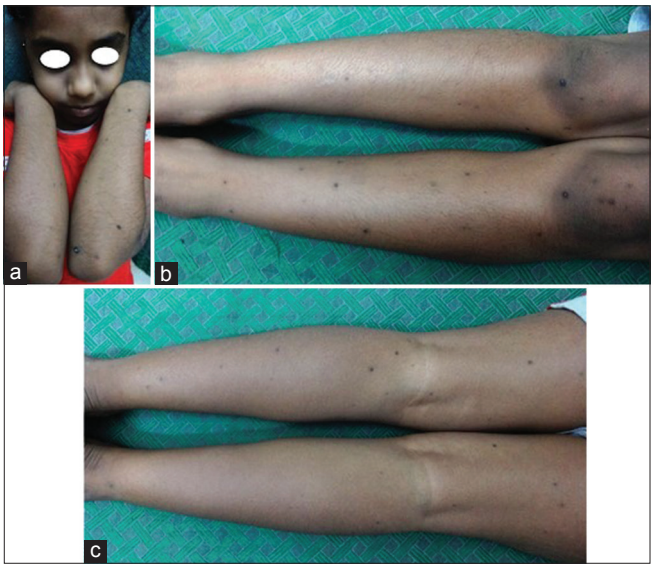

Figure 4: (a) Multiple comedones with pigmented papular lesions more clustered over Elbows and facial sparing (b and c) multiple comedonal papules over lower limbs

asymptomatic, pruritus[6,8,9] and burning sensation[5] may accompany those lesions on the scrotum and penis. A few of the keratotic papules form painful, inflamed swellings over the back.[4]

This entity has distinctive histopathology showing crater-like invagination filled with lamellar keratinous material with foci of dyskeratosis. Acantholysis may also be seen sometimes.[6,7] Electron microscopy reveals decreased number of desmosomal attachments within the stratum malpighii.[6] Recently, a new entity of "familial disseminated comedones without dyskeratosis" has been proposed for a group of family members with a similar presentation and histopathology showing no dyskeratosis.[3]

Differential diagnosis includes Kyrle's disease, reactive perforating collagenosis, keratosis pilaris, perforating folliculitis, nevus comedonicus, and acne vulgaris. Familial comedonal Darier disease is a close differential diagnosis which presents with follicular and extrafollicular greasy, hyperkeratotic papules and plaques in seborrheic areas. Histopathology of Darier's disease shows dyskeratosis with corps ronds and grains, suprabasal acantholysis and villi, which are diagnostic.[11] Nevus comedonicus, which develops shortly after birth or before the age of 10 years, presents with closely arranged, dilated follicular openings with keratinous plugs resembling classical comedones predominantly over face and neck; mostly unilateral, although it can have bilateral, linear, interrupted, segmental, or Blaschkoid distribution. The classical clinical features, positive family history, and histopathology confirmed diagnosis in our case.

Management is difficult task. Various treatment modalities including topical retinoids and oral isotretinoin have proved ineffective.[4,6,7,10] However, frequent sun exposure[5] and carbon dioxide laser[7,8] have shown promising results. Although in FDC lesions are asymptomatic, involvement of face is very distressing increasing the psychological disability of the patients. Further studies might explore the molecular pathology, which may widen our gaze regarding the better treatment modalities.

What's new?

Our case is peculiar with respect to early age of onset of lesions in her 9-year-old daughter, with sparing of face till now. Although FDC is not infrequent, we project this case to minimize underreporting and to enhance the awareness for larger studies to unleash the exact pathogenesis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Rodin HH, Blakenship ML, Bernstein G. Diffuse familial comedones. Arch Dermatol 1967;96:145-6.
2. Carneiro SJ, Dickson JE, Knox JM. Familial dyskeratotic comedones. Arch Dermatol 1972;105:249-51.
3. Cheng MJ, Chen WC, Happle R, Song ZQ. Familial disseminated comedones without dyskeratosis: Report of an affected family and review of the literature. Dermatology 2014;228:303-6.
4. Kumaran MS, Appachu D, Jayaseelan E. Familial dyskeratotic comedones. Indian J Dermatol Venereol Leprol 2008;74:142-4.
5. Leppard, BJ. Familial dyskeratotic comedones.Clin Exp Derm 1982;7:329-32.
6. Hall JR, Holder W, Knox JM, Verani R. Familial dyskeratotic comedones. A report of three cases and review of the literature. J Am Acad Dermatol 1987;17:808-14.
7. Hallermann C, Bertsch HP. Two sisters with familial dyskeratotic comedones. Eur J Dermatol 2004,14:214-5.
8. Stander S Rutten A, Mete D. Familial dyskeratotic comedones: A rare entity. Hautarzt 2001;52:533-6.
9. Van Geel NA, Kockaert M, Neumann HA. Familial dyskeratotic comedones. Br J Dermatol 1999;140:956-9.
10. Michot C, Guilhou JJ, Bessis D. Familial dyskeratotic comedones. Ann Dermatol Venerol 2004;131:811-3.
11. Chung J, Kim JY, Gye J, Namkoong S, Hong SP, Park BC, *et al*. A case of familial comedonal Darier's disease. Ann Dermatol 2011;23(Suppl 3):S398-401.